The team of people involved can include a doctor specialising in the care of children (a paediatrician), your GP, a heart specialist (cardiologist), an eye specialist (ophthalmologist), a speech and language specialist, a physiotherapist, an occupational therapist and/or a social worker.

Your child will have regular medical follow-ups with his/her team throughout their life to monitor for any problems that may arise.

It is also important to maintain a healthy, balanced diet and regular exercise to reduce the risk of weight gain and obesity.

What is the outlook (prognosis) for Trisomy 21?

Due to advancements in medical treatments at least half of the people diagnosed with Trisomy 21 now live into their 50's and 60's. Congenital heart problems are often the cause of death in those who do not survive.

Support for parents and carers

Great support and advice is available for parents dealing with a pregnancy diagnosed with Trisomy 21 and you should never feel that you are alone.

You will be able to access pregnancy and genetic counselling through your Fetal Medicine Unit for you and your whanau.

You may also like to contact:

www.adsa.org.nz

The Auckland Down Syndrome Association Inc ("ADSA") is an organisation that provides information and support for people with Trisomy 21 and their families.

www.parent2parent.org.nz/

Parent to Parent New Zealand is an information and support network for parents of children with special needs ranging from the very common to the most rare conditions.

community.upsideofdown.co.nz

Support for friends and family of people with Trisomy 21 and other special needs.

References

http://www.britannica.com

TRISOMY 21 leaflet.doc

TRISOMY 21 leaflet.doc

http://www.nzdsa.org.nz

For more information please contact your local NZMFMN Unit

Auckland: 09 307 4949 ext 24951

Wellington: 04 806 0774

Christchurch: 03 364 4557

New Zealand Maternal Fetal Medicine Network

NZMFMN@adhb.govt.nz

Trisomy 21 (Down Syndrome)

New Zealand Maternal Fetal Medicine Network

Reviewed: October 2015

What is a chromosome?

Inside our cells there are microscopic, threadlike parts called chromosomes. They carry hereditary information from one generation to the next in the form of genes. A chromosome is made up of protein and deoxyribonucleic acid (DNA) which determines a person's inherited traits such as eye colour or left/right handedness.

You inherit 23 chromosomes from your mother and 23 from your father for a total of 46 chromosomes arranged in pairs (one from each parent). They are numbered in pairs from 1 to 22. The last pair (23) is the sex chromosomes and determine if the developing baby is a boy or a girl.

What is Trisomy 21?

Trisomy 21 results when there are three copies of chromosome 21 instead of the usual two.

The extra genetic material disrupts the normal course of development in the fetus and this causes the features typical of the condition. All people with Trisomy 21 have some degree of learning disability. They are also at increased risk of having various other medical problems. It cannot be cured but treatment and support can help someone with Trisomy 21 to lead an active life.

How common is Trisomy 21?

"In New Zealand one baby in about 1000 is born with Trisomy 21; that is, one or more babies with Trisomy 21 born every week" (www.adsa.org.nz).

How is Trisomy 21 diagnosed?

If you have come to our Fetal Medicine Unit it is likely that you have already had the screening test for Trisomy 21 (MSS1 or MSS2) and that it shows an increased risk.

It is important to understand that this screening test does not give a definitive answer as to whether your baby does have Trisomy 21- it just gives a risk.

If your screening test shows an increased risk there is a test available in the private sector called a non-invasive (blood test) prenatal test (NIPT). It is currently not publicly funded and will cost you approximately $1000. If this test was low risk it would virtually exclude the risk of Trisomy 21, however, if it was positive you would still be offered invasive testing for a definitive answer.

There are two invasive antenatal diagnostic tests that can be done- an amniocentesis and chorionic villus sampling (CVS).

These tests carry a small risk of miscarriage (approximately 0.1% for amniocentesis and 0.2% for CVS). An amniocentesis/CVS allows us to detect chromosomal abnormalities from a sample of the fluid from around your baby or from a sample of tissue from the placenta. It is obtained by inserting a thin needle through your abdomen and into your uterus.

Features

There are certain features that are common in people with Trisomy 21; however, not everyone will have all features. Some people may only be mildly affected whilst others more severely. We are unable to tell how affected your child will be until after he/she is born.

Facial features

- Oval-shaped face
- Eyes that slant upwards and have an extra fold of skin on the upper eyelid known as an epicanthal fold.
- Small mouth and protruding tongue
- Flatter head shape
- Small nose with a low flat bridge
- Small low-set ears

Other physical features

- Single palmar crease
- Short fingers and a little finger that curves inwards
- Joints that seem 'looser' than normal
- Sandal gap deformity (gap between the 1st and 2nd toes
- Reduced muscle tone
- Low birth weight

Heart defects

Children with Trisomy 21 are much more likely to also suffer from congenital (born with) heart disease. Your baby has an approximately 50% chance of having a problem with his/her heart.

The Fetal Medicine team will schedule an echocardiogram in conjunction with your anatomy scan. This will either be in Auckland, Wellington or Christchurch depending on where you live.

The most common heart problems are:

- Atrioventricular Septal Defects (most common)
- Ventricular Septal Defects
- Atrial Septal Defects

(These are more commonly known as 'holes in the heart')

- Patent Ductus Arteriosus

(This is a heart condition where abnormal blood flow occurs between two of the major arteries connected to the heart)

- Tetralogy of Fallot

(A heart condition that involves four different structural abnormalities)

Typical behaviour

People with Trisomy 21 tend to be naturally very warm and cheerful and quite spontaneous in their behaviour, however, they are also at a greater risk for depression and anxiety disorders.

Other problems

Someone with Trisomy 21 is also at increased risk of developing other medical problems or conditions.

These can include:

- Vision problems
- Hearing problems
- Slower growth resulting in short stature as adults
- Leukaemia
- Obesity can often develop during adolescence
- Poor immune function
- Thyroid disorders
- Diabetes.
- Dementia and other forms of mental illness
- Epilepsy
- Reduced fertility
- Duodenal atresia
- Hirschsprung's disease

Care

With great support and understanding from whanau, friends and the wider community, someone with Trisomy 21 can lead a very active and fulfilling life.

Early intervention programmes such as speech and language therapy and physiotherapy from birth are important. The earlier these programmes are implemented the better the long-term outcome is likely to be and the more likely your child is to reach his/her full potential.

A number of different healthcare providers are likely to be involved in your child's care.